



GIG
CYMRU
NHS
WALES

Rhwydwaith Gweithredu
Clefydau Prin
Rare Diseases
Implementation Network

Rare Disease Research Network for Wales Launch

Rare Disease Implementation Network

15th January 2025



GIG
CYMRU
NHS
WALES

Y Weithrediaeth
Executive

Agenda



**Rare Diseases Implementation Network
Rare Disease Research Network (RDRN) launch
Wednesday 15th January 2025 – 12.30 – 13.30**

[Click here to join the meeting](#)

Agenda

	Item No.	Item	Paper/ Verbal	Lead
12:30	1	Welcome and RD Action plan	Verbal	Dr Jamie Duckers Rare Disease Clinical lead
12:35	2	Update on current Rare Disease research in Wales and UK Rare Disease Landscape report	Presentation Paper 1 and 2 (reports UK and Wales)	Dr Francis Sansbury HCRW Rare Disease clinical lead
12:50	3	The importance of Research in Rare Diseases – A CF story	Verbal	Dr Jamie Duckers Rare Disease Clinical Lead
13:00	4	Life Science Hub (LSH) Wales, support available for researchers	Presentation	Elizabeth Rees Funding Programme Lead -LSH
13:20	5	Ask the audience for suggested topics at next meeting and what they want from the network	Verbal	All
14:30	6	Close		

The RDRL (rare disease research landscape) project: a Wales perspective

Francis Sansbury

HCRW (Health and Care Research Wales) lead for genomics and rare disease

15th January 2025

What is an HCRW specialty lead?

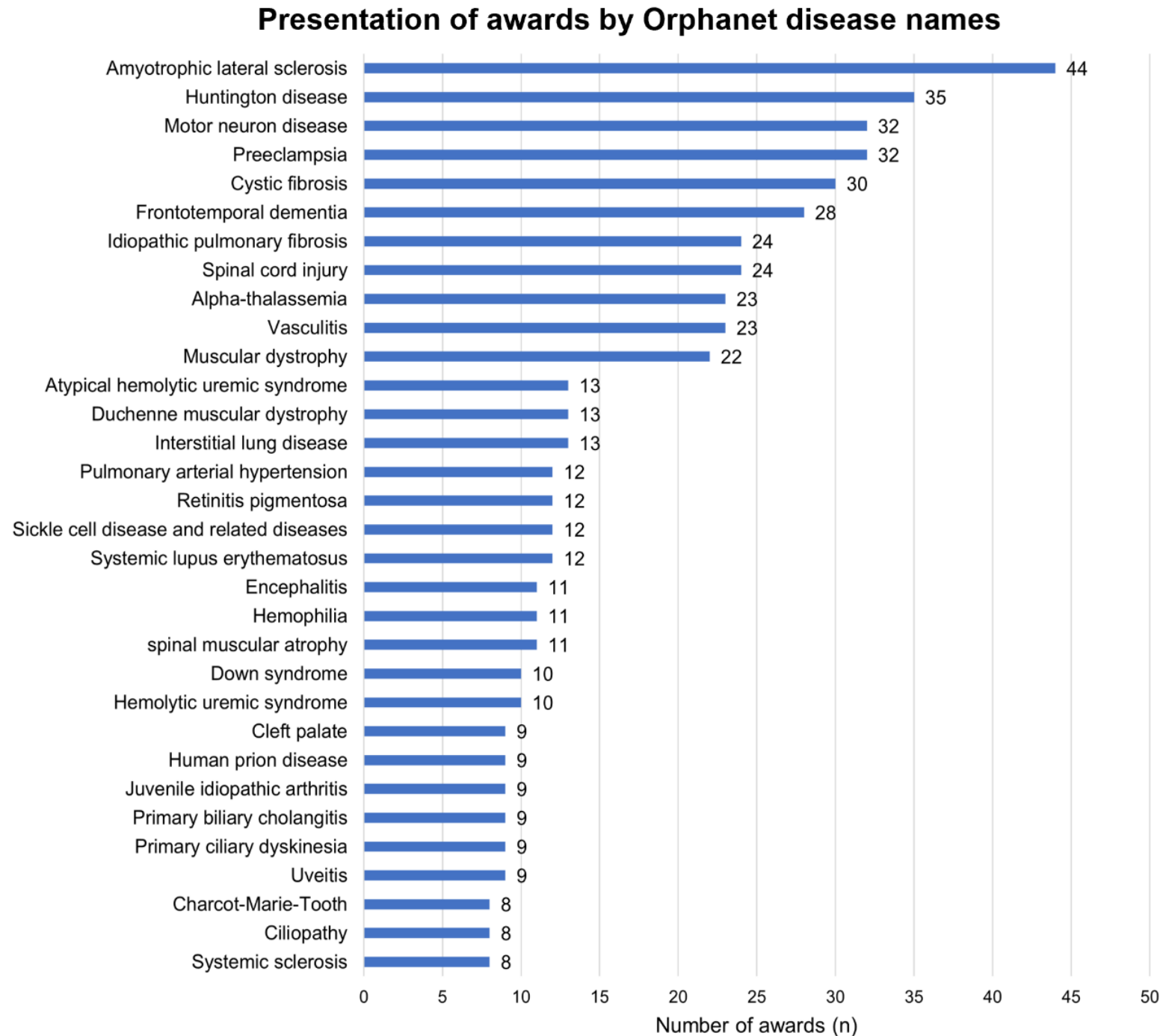
- 1 session / week (rest of week: consultant clinical geneticist)
 - Provide important strategic support as part of the Support & Delivery Service
 - Engages and supports a network of researchers within the relevant speciality area [i.e. GARD (Genomics and Rare Disease) – others do neuro / respiratory etc.]
 - Encourage NHS participation in portfolio studies (for GARD)
 - Support the successful set-up and delivery of research studies across Wales
 - Support a culture of research within health and social care settings where relevant
 - Be an active member of the relevant NIHR Specialty Group
 - Genetic NSG (national specialty group) abolished as of last October
 - (Not actually to do one's own research)
- For me: also represent Wales on RDRL (rare disease research landscape) project steering group

UK RDRL project

- Two phases
 - First phase report in autumn 2023 (NIHR Open)
 - Mapped RD (rare disease) research across the UK
 - Second phase underway
 - Aim: identify research gaps and priorities that can be used to inform rare diseases research strategy in the future
 - Two steering group meetings so far
 - Priority-setting workshop in autumn in London – awaiting results of that
- UK RDRL Phase 1 report (on NIHR Open)
- Life Sciences Hub Wales: RDRL in Wales

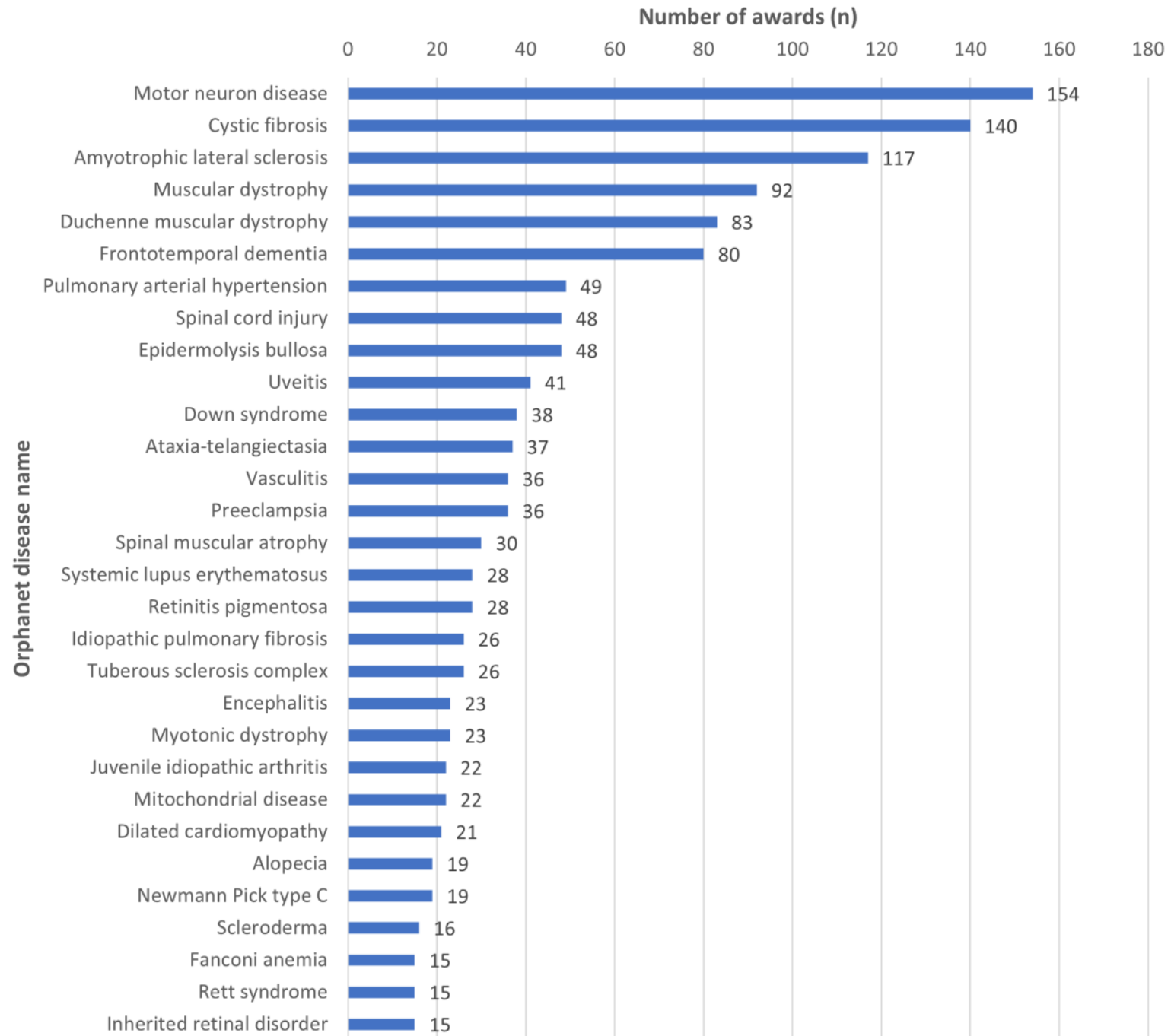
UK Phase 1 results

- NIHR & MRC RD portfolio
- Awards / disease
 - Neurological predominates then respiratory



UK Phase 1 results

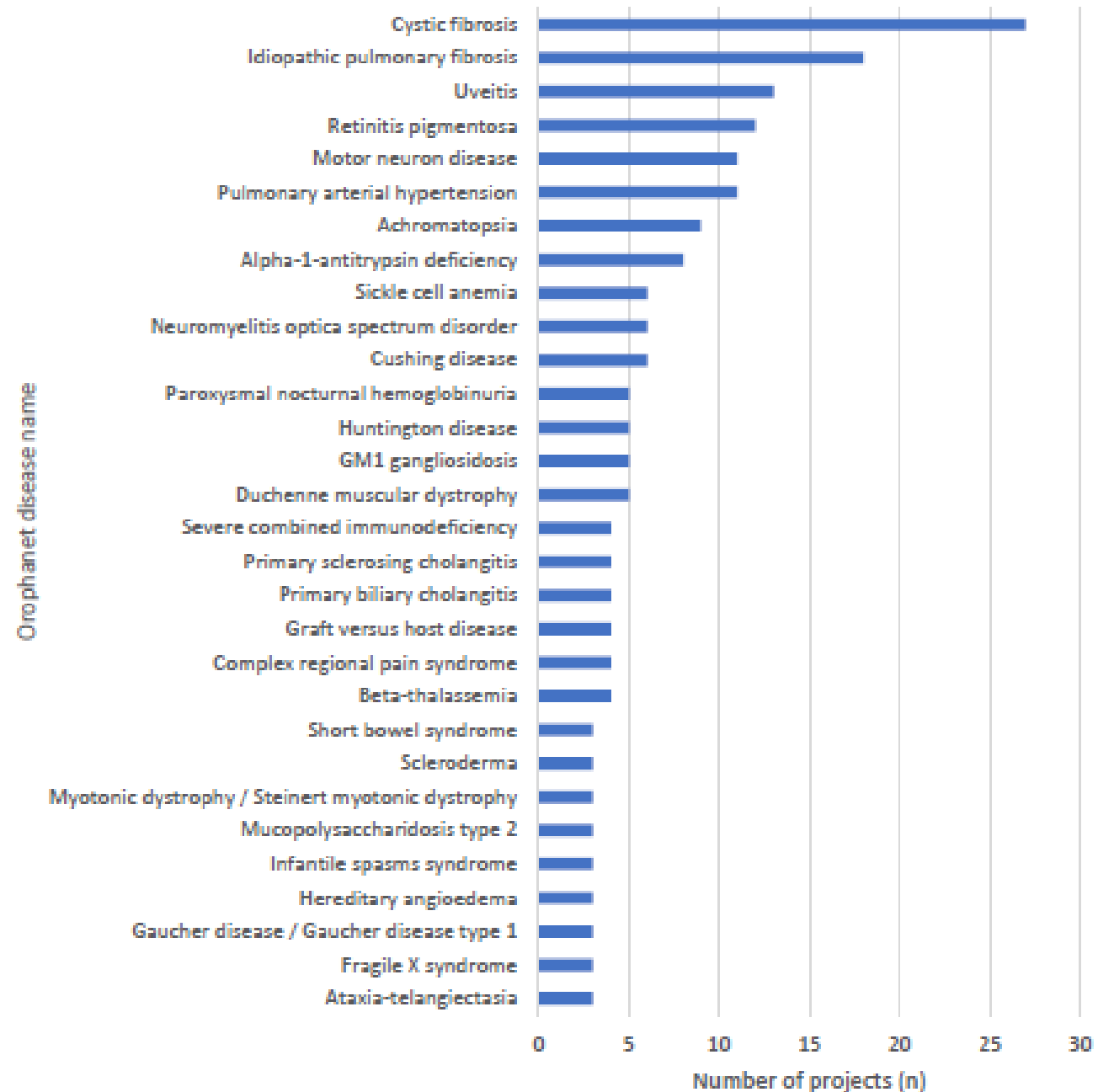
- AMRC (Association of Medical Research Charities) portfolio
- Awards / disease
 - Neurological and respiratory predominate
 - Are cancers hiding in the tail?



UK Phase 1 results

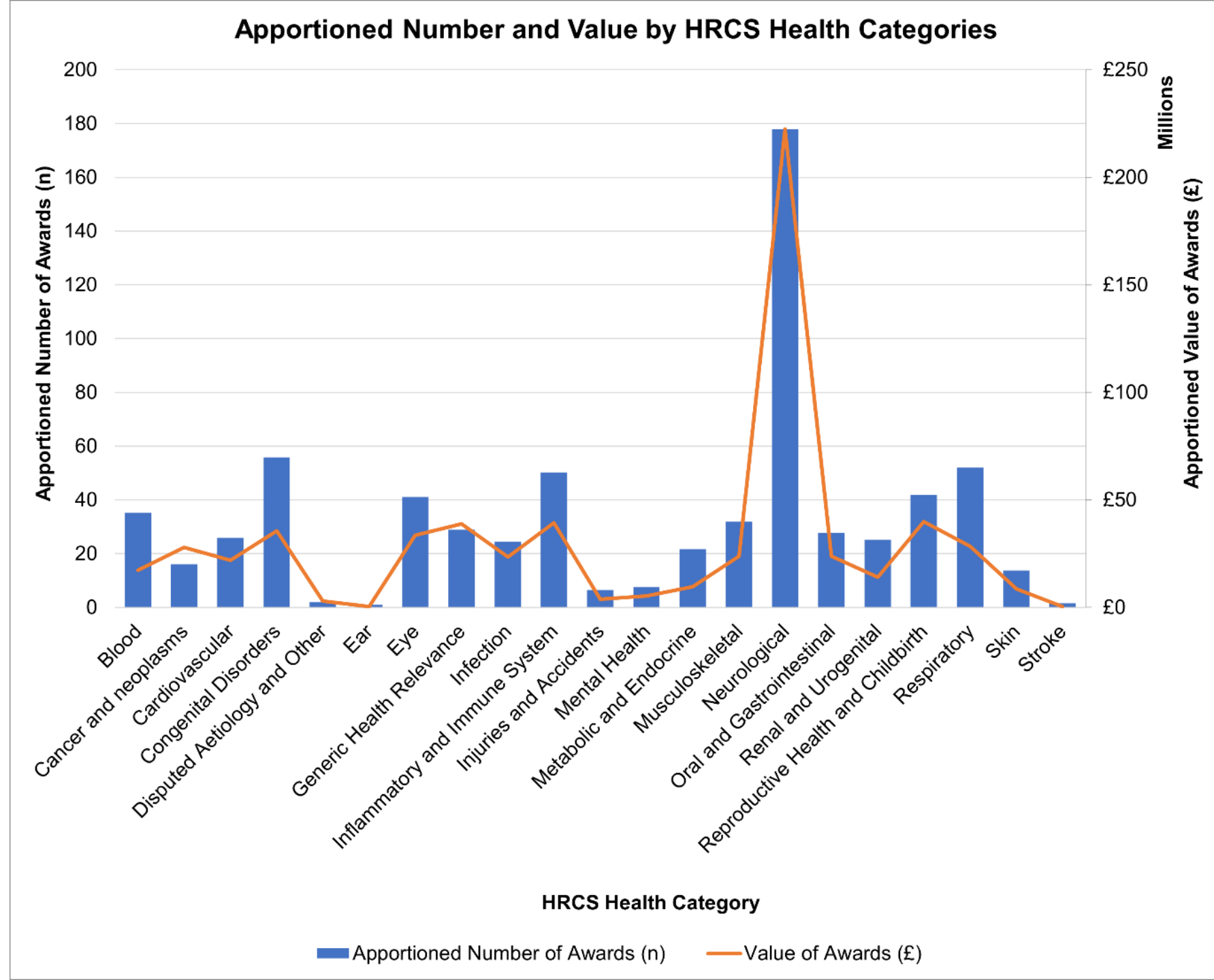
- Industry portfolio
- Awards / disease
 - Respiratory predominates

Number of industry projects by Orphanet disease names



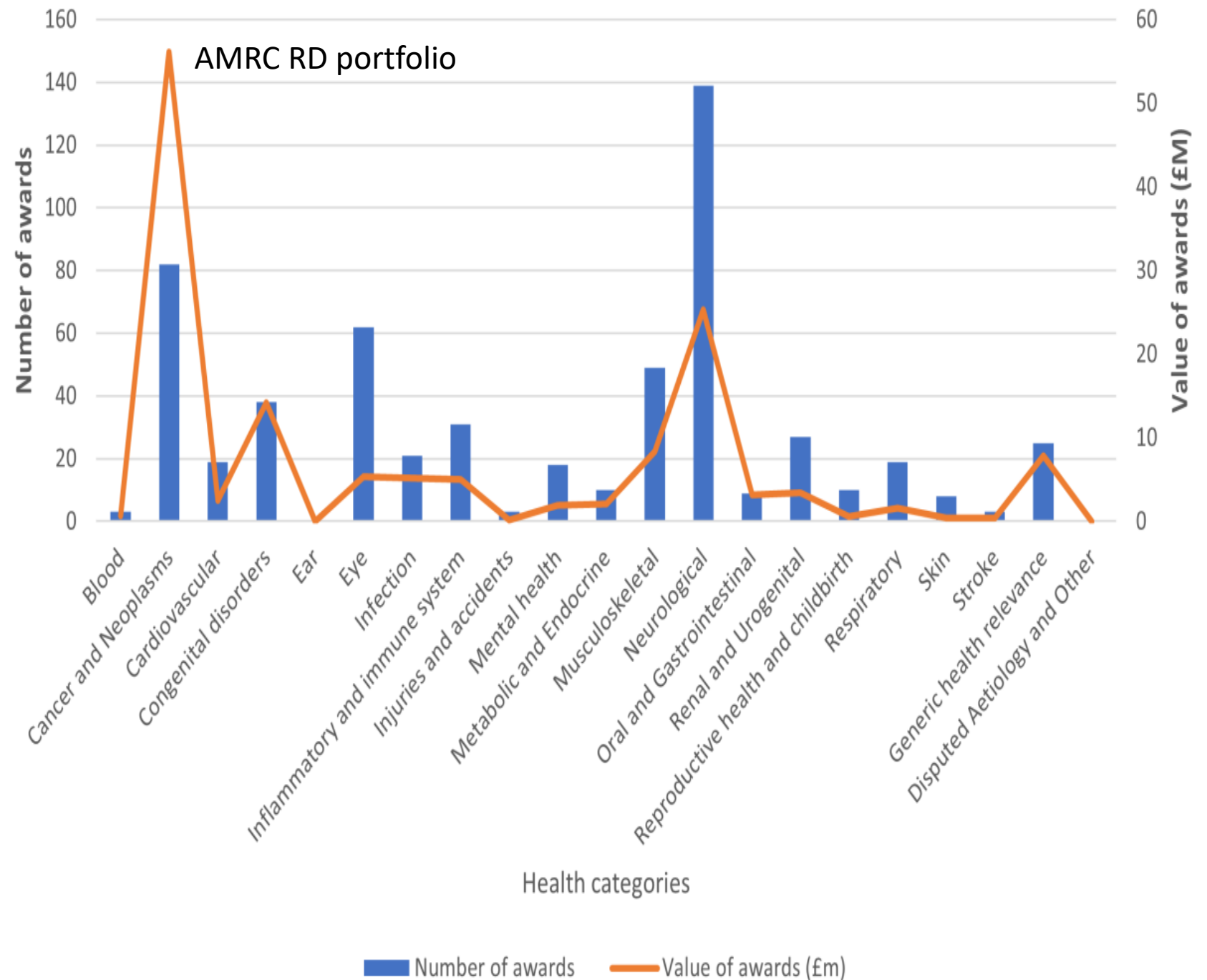
Phase 1 results

- NIHR & MRC RD portfolio
 - Neurological predominates



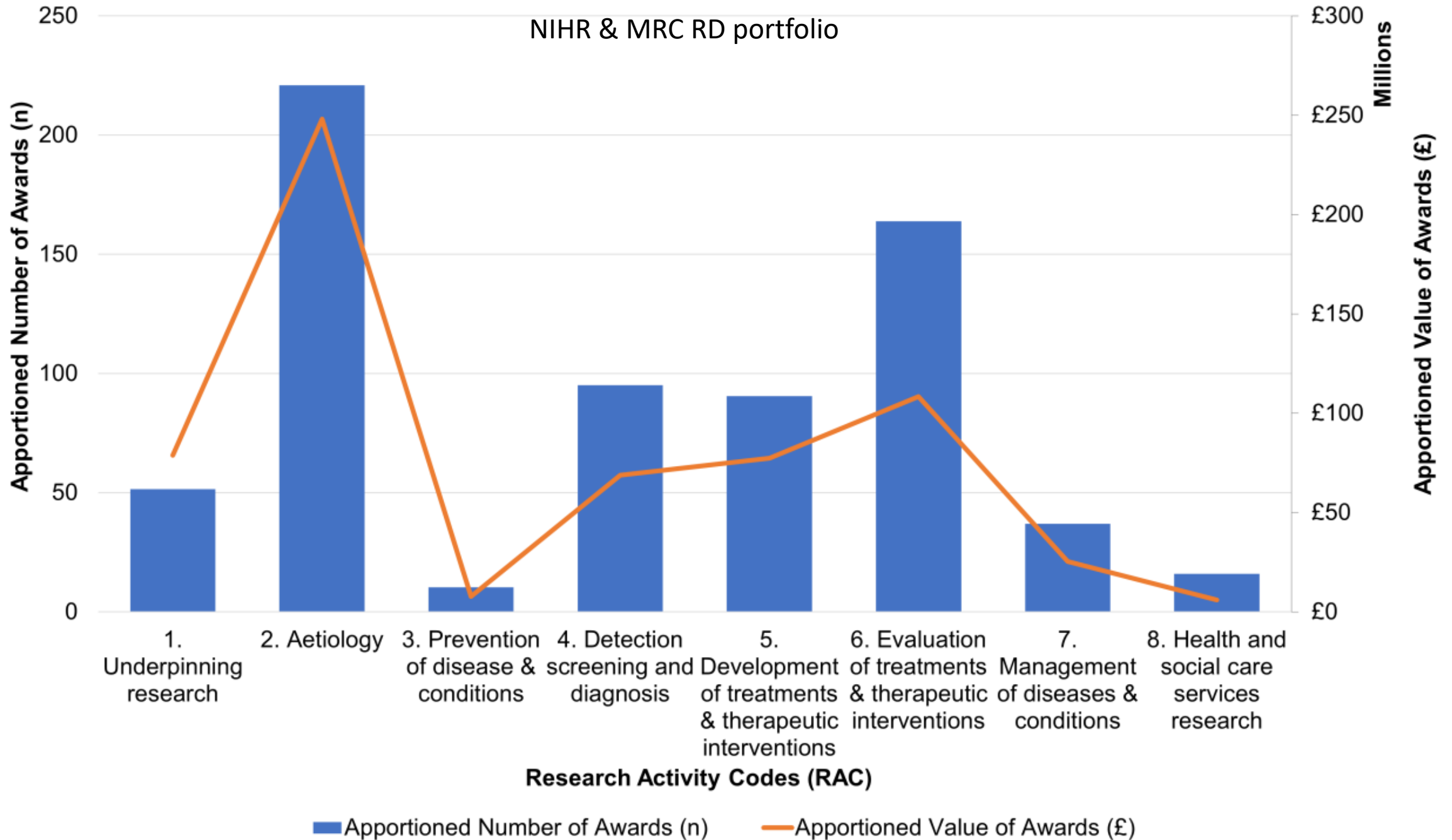
Phase 1 results

- AMRC (Association of Medical Research Charities)
 - Neurological and cancer
 - CRUK (Cancer Research UK)?

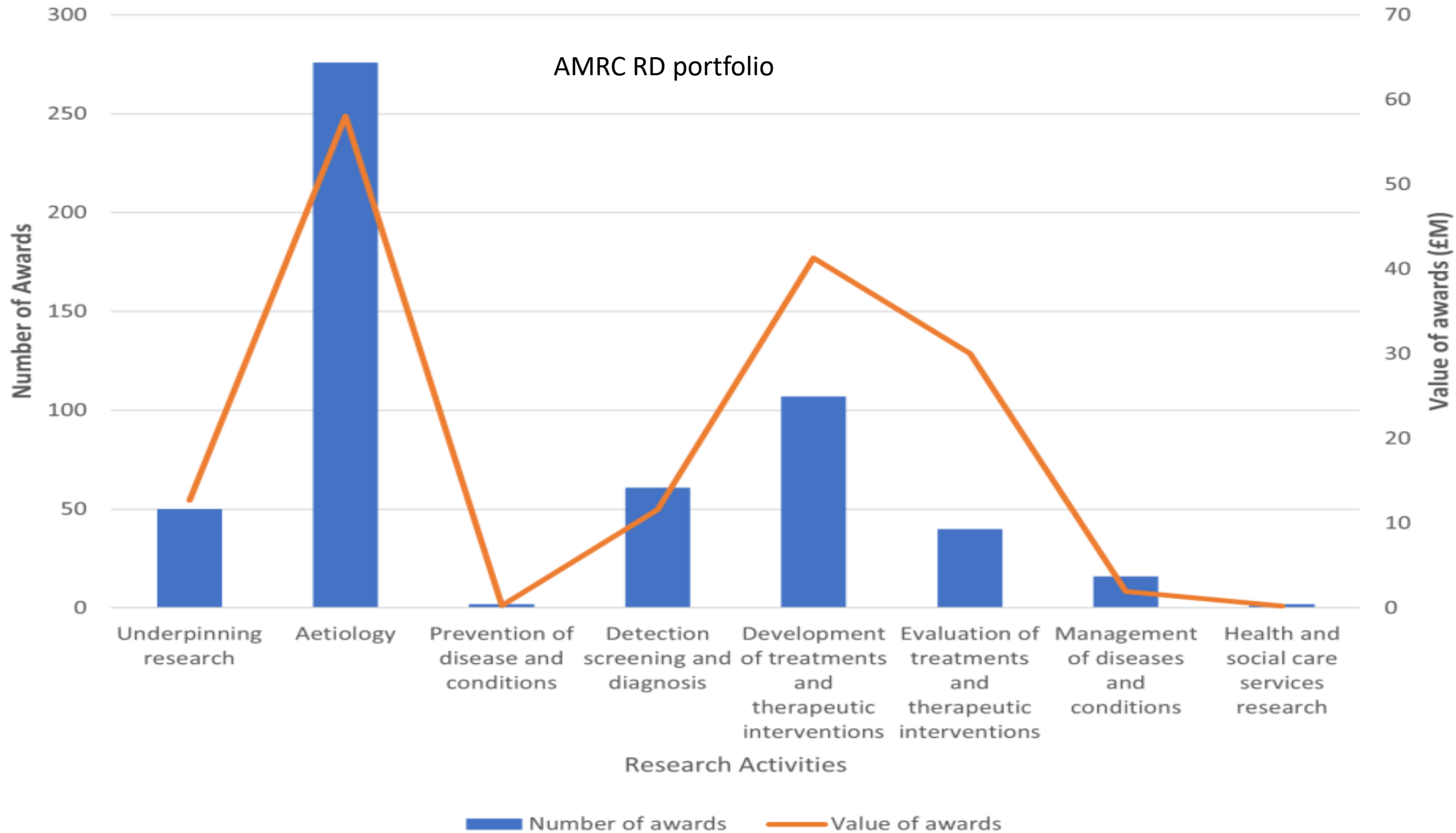


Apportioned Number and Value by HRCS Research Activity Codes

NIHR & MRC RD portfolio



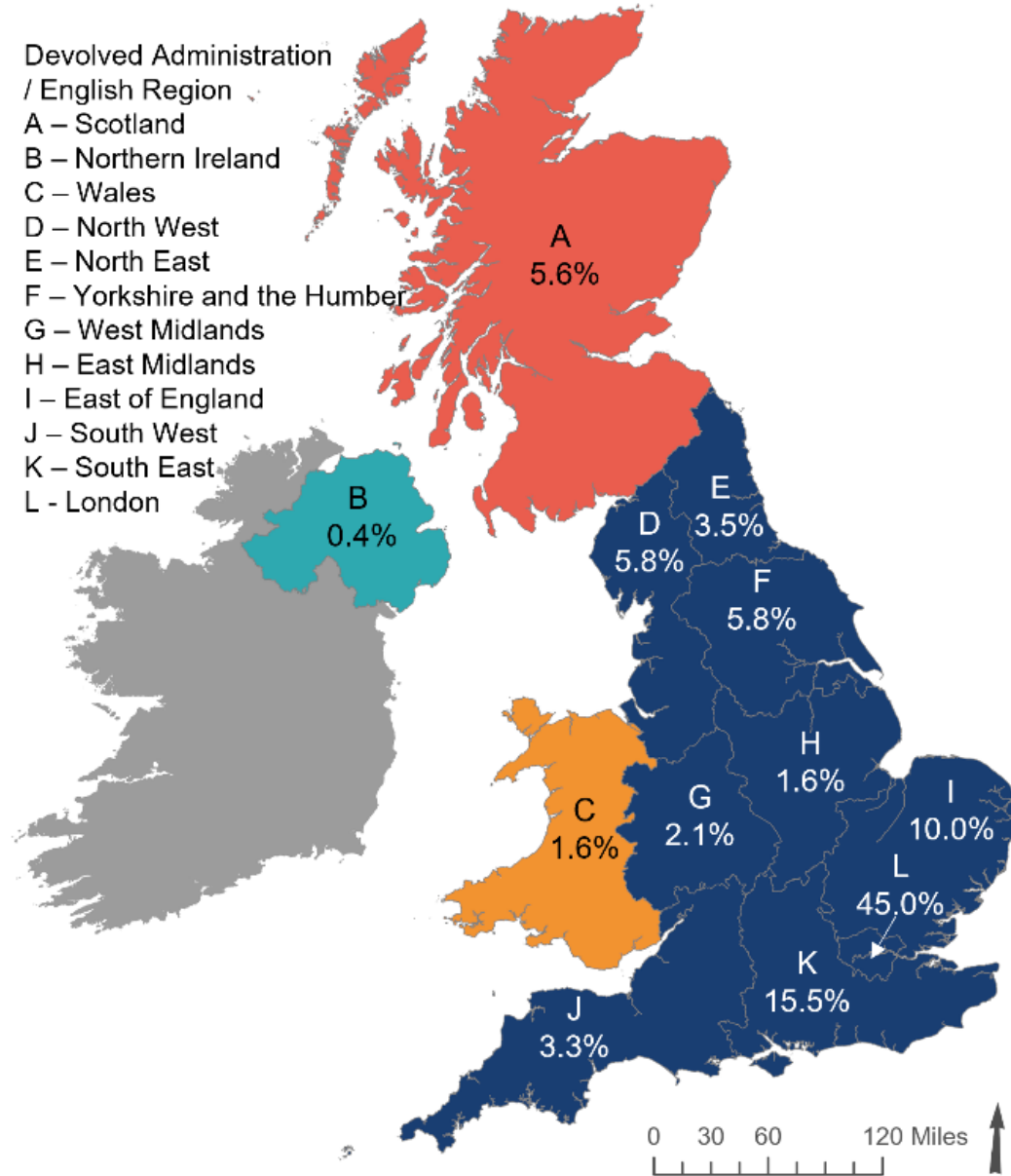
AMRC RD portfolio



MRC and NIHR Programmes Rare Disease Combined Portfolio - Distribution of Funding Across the UK

Devolved Administration / English Region

- A – Scotland
- B – Northern Ireland
- C – Wales
- D – North West
- E – North East
- F – Yorkshire and the Humber
- G – West Midlands
- H – East Midlands
- I – East of England
- J – South West
- K – South East
- L - London

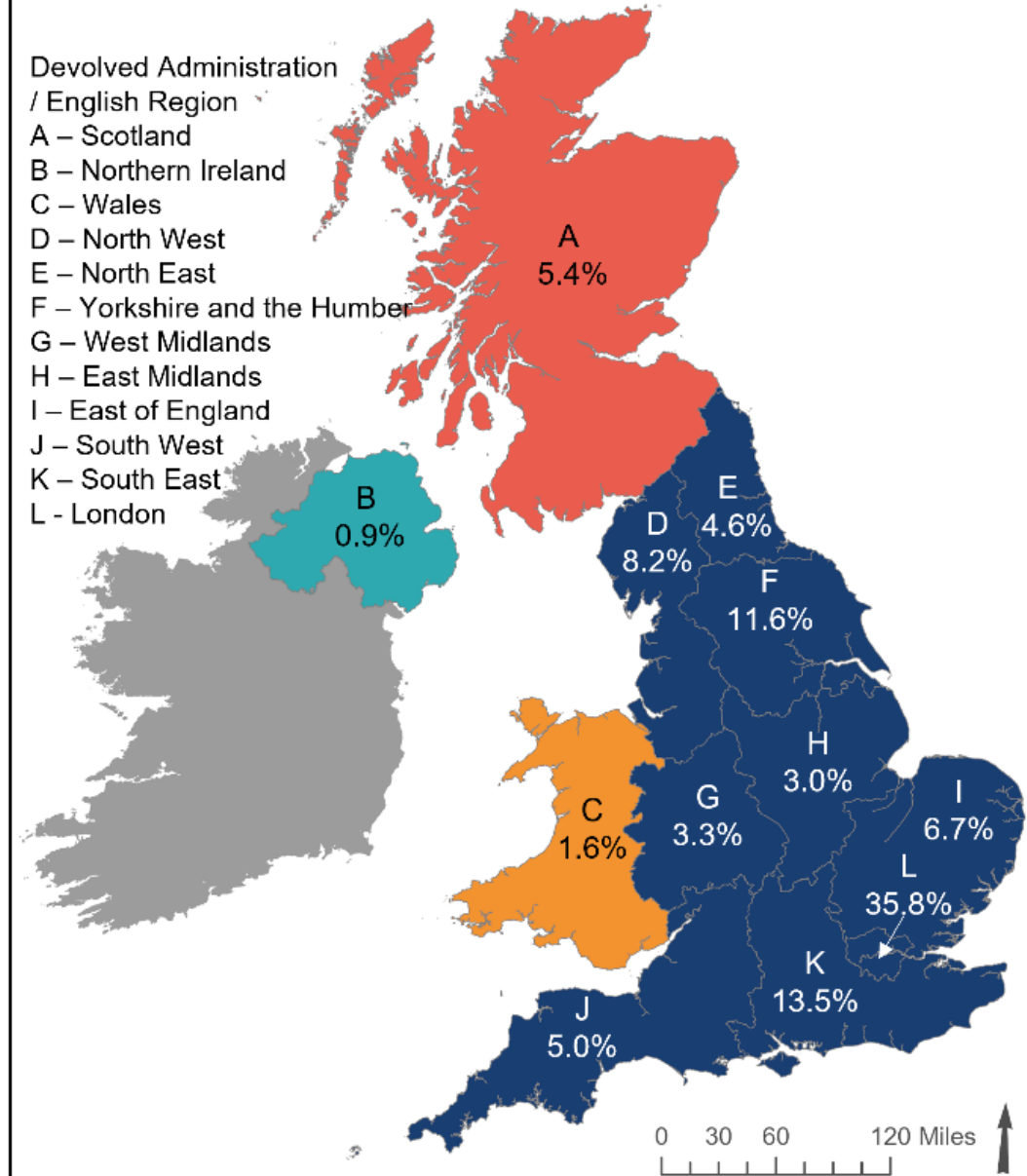


Sources: MRC and NIHR - 3 awards excluded as Research Organisation outside the UK
 Contains OS data © Crown copyright and database right [2023]
 Sources: Office for National Statistics licensed under the Open Government Licence v.3.0
 Prepared by the NIHR Centre for Business Intelligence

MRC and NIHR Programmes Rare Disease Combined Portfolio - Distribution of Awards Across the UK

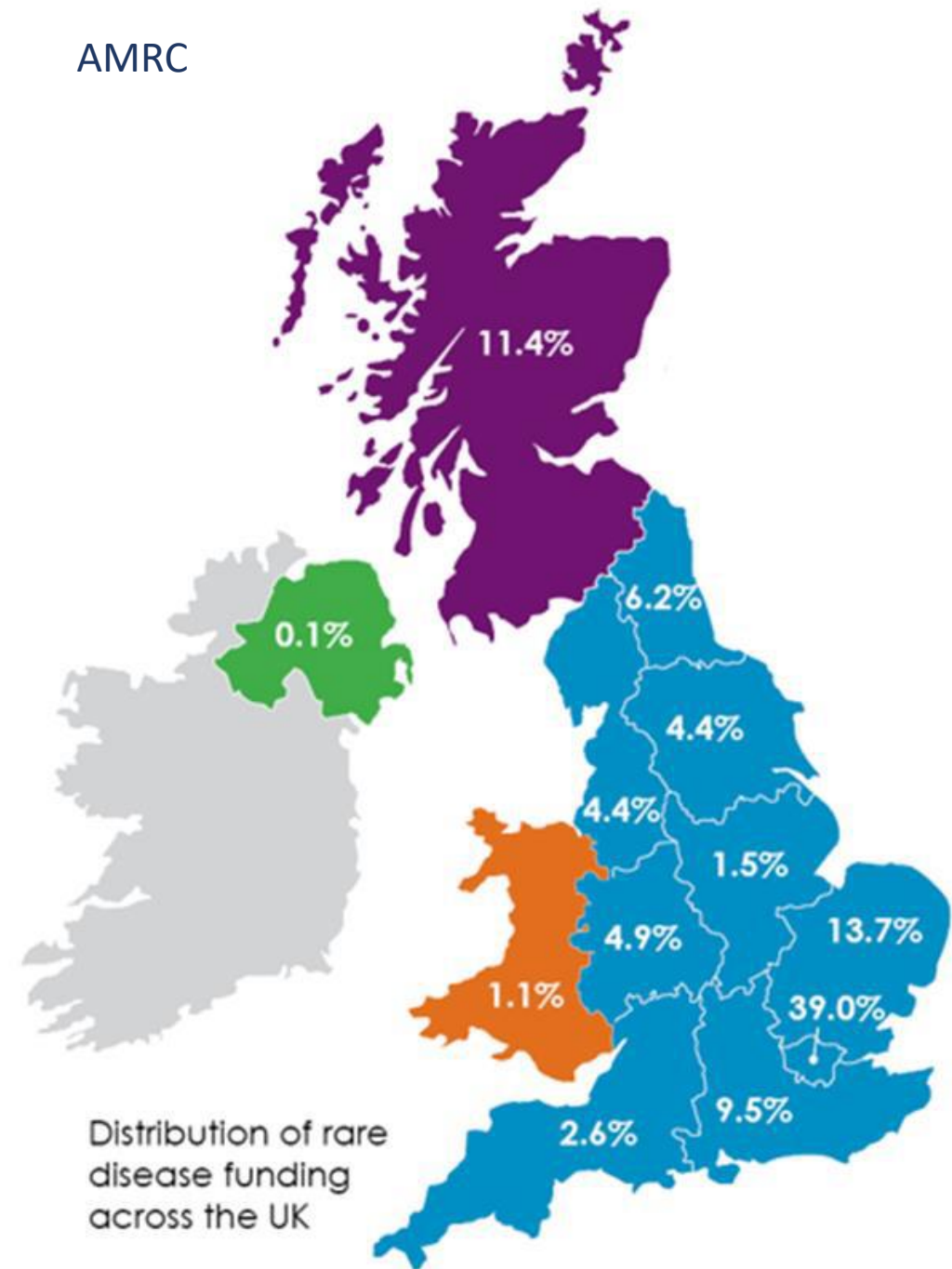
Devolved Administration / English Region

- A – Scotland
- B – Northern Ireland
- C – Wales
- D – North West
- E – North East
- F – Yorkshire and the Humber
- G – West Midlands
- H – East Midlands
- I – East of England
- J – South West
- K – South East
- L - London



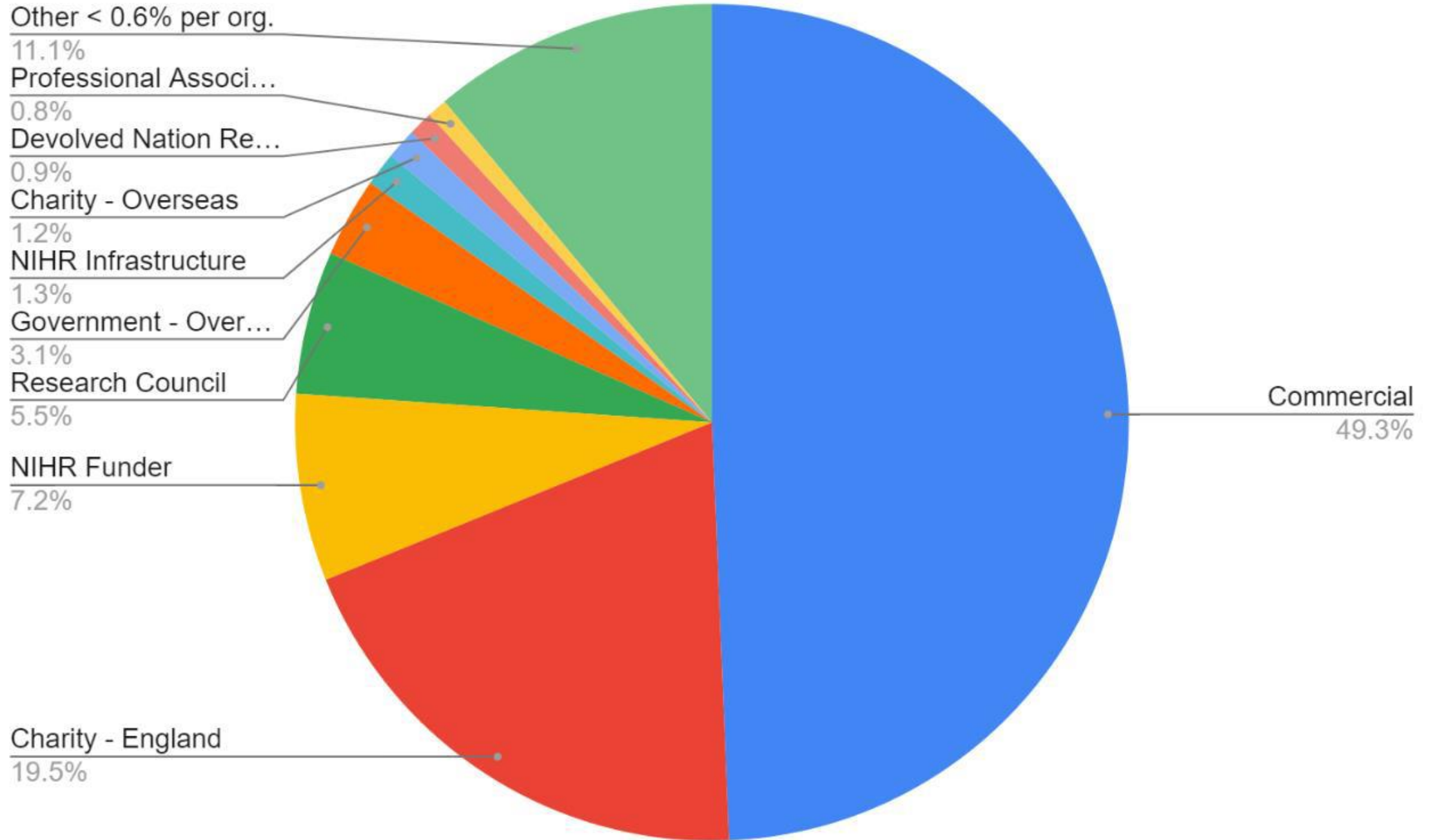
Sources: MRC and NIHR - 3 awards excluded as Research Organisation outside the UK
 Contains OS data © Crown copyright and database right [2023]
 Sources: Office for National Statistics licensed under the Open Government Licence v.3.0
 Prepared by the NIHR Centre for Business Intelligence

AMRC

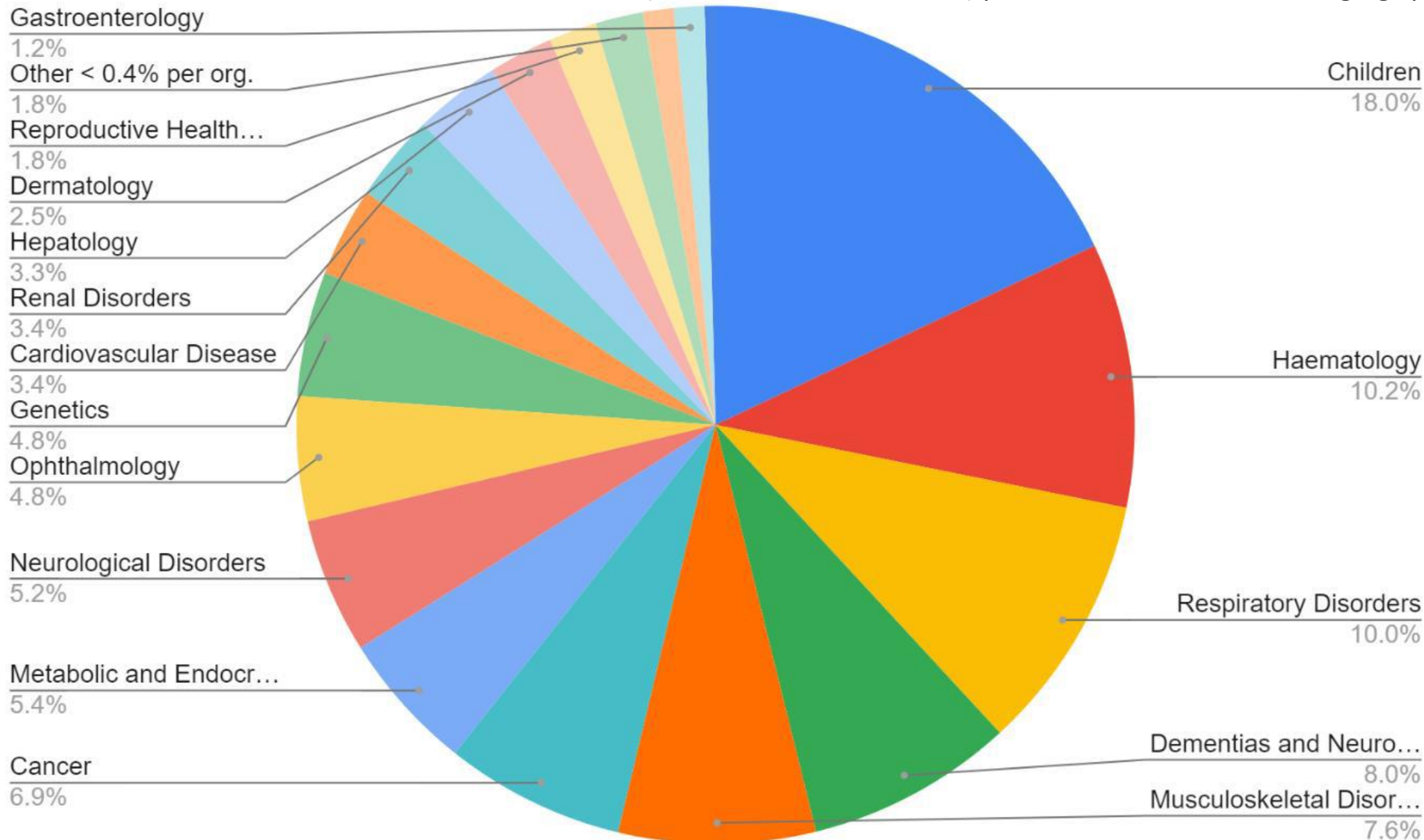


Distribution of rare disease funding across the UK

RD studies on NIHR CRN (clinical research network) portfolio 2016-21: % / type of funder

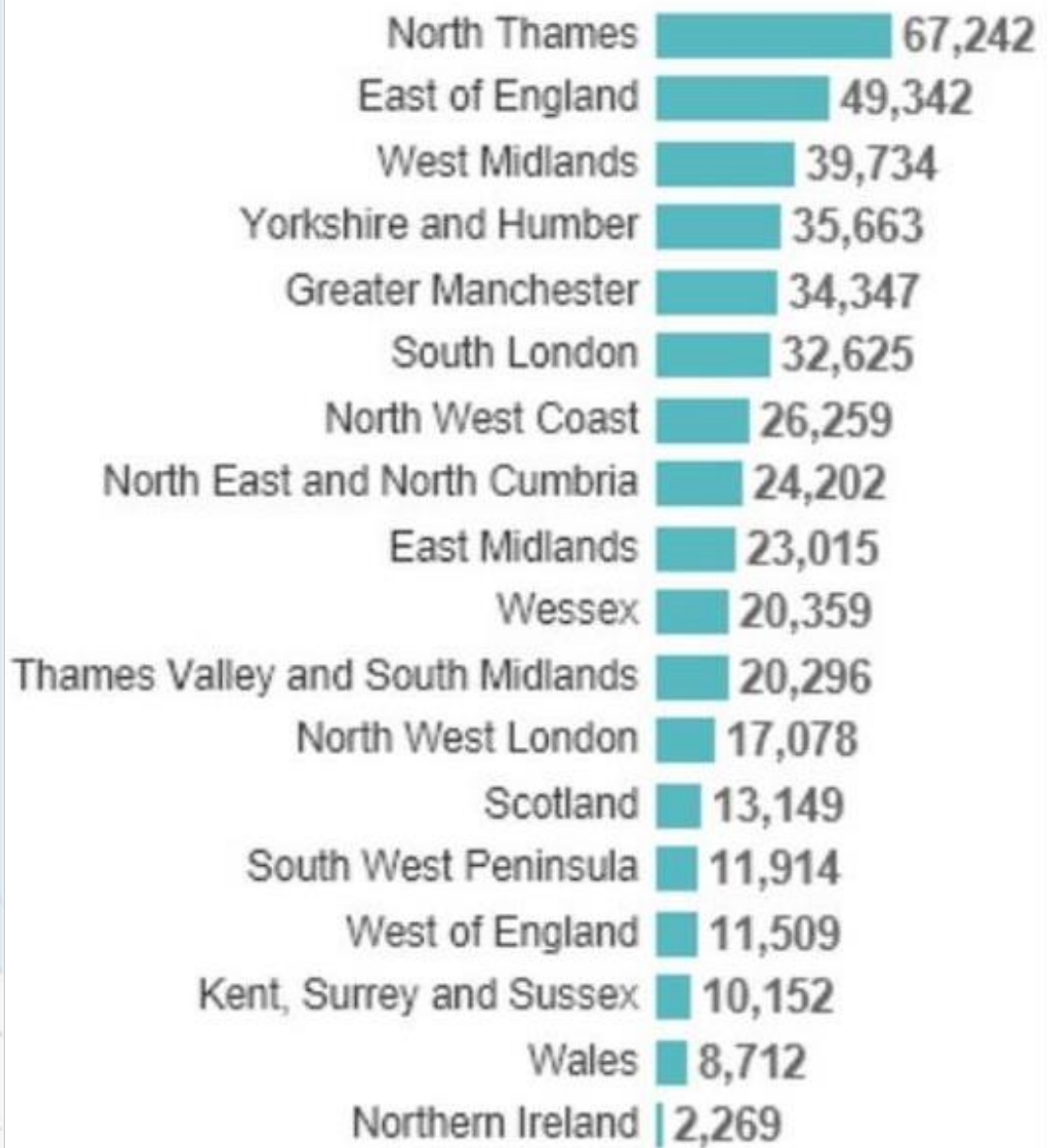


RD studies on NIHR CRN (clinical research network) portfolio 2016-21: % / managing specialty





Numbers of participants 2016-21 by CRN (clinical research network)



Wales and NI lowest proportion of funding

- If were per capita UK, expect Wales 4.6 %
 - Population mid 2023: 3164000 / 68265209 in UK
- Only 1 NIHR / MRC career development award in Wales
- Data collection predates UK Rare Disease Research Platform (MRC and NIHR), and LifeArc Rare Disease Translational Challenge
 - E.g. does not include Swansea metabolomics award
- Snapshot in time
 - E.g. section on WGP (Wales Gene Park) out of date

Wales report

- Comparison of UK total and Wales data from MRC and NIHR Programmes Rare Disease Combined Portfolio 2016-21

	Total award value	No. awards	Award value per annum	Median award value
Total UK dataset	£ 626,918,074	698	£ 125,383,615	£ 425,065
Wales	£ 9,785,066	11	£ 1,957,013	£ 687,871
Wales as a % of UK total	1.6 %	1.6 %	1.6 %	

- Neuro [especially dementia / HD (Huntington disease)]
- Paediatrics (respiratory)
- Cardiff
- £1 M for 100000 genomes project
 - Not research funding in England

#	Funder Programme	Title	Research Organisation	Awarding body	Award Value
1	Health Technology Assessment	AZithromycin Therapy for Chronic lung disease (AZTEC): A randomised, placebo controlled trial of azithromycin for the prevention of chronic lung disease of prematurity in preterm infants	Cardiff University	NIHR	£2,361,018
2	MRC SIEBEL	Cell therapy for Huntington's disease: addressing critical knowledge gaps	Cardiff University	MRC	£1,996,455
3	MRC SIEBEL	LIMK1 inhibitors - A novel, disease-modifying approach for the treatment of fragile X syndrome	Cardiff University	MRC	£1,980,780
4	MRC LIS	The Wales Genomic Medicine Centre	Cardiff University	MRC	£1,000,000
5	MRC SIEBEL	How CTIP2 deficiency drives medium spiny neuron degeneration and dysfunction: implications in Huntington's disease pathogenesis	Cardiff University	MRC	£708,068
6	MRC LIS	(Epi)genome editing to combat expanded CAG/CTG repeat disorders	UK Dementia Research Institute at Cardiff University	MRC	£687,871
7	MRC LIS	Understanding the role of neurodegenerative disease risk genes in the ageing nervous system	Cardiff University	MRC	£485,234
8*	MRC SIEBEL	Understanding the role of DNA repair in Huntington's Disease pathogenesis: towards new therapeutic targets	Cardiff University	MRC	£309,744
9	MRC SIEBEL	Cardiff Fetal Tissue Bank: Quality assured tissue for biomedical research and clinical trial in neurodegenerative disease	Cardiff University	MRC	£202,304
10	MRC LIS	Exploring the proteome of CAG/CTG repeat expansions	UK Dementia Research Institute at Cardiff University	MRC	£38,733
11	MRC SIEBEL	Investigating the effects of CAG repeat structure and MSH3 variants on the molecular biology of Huntington's disease	Cardiff University	MRC	£14,859

UK RDRL phase 2: main issue from my perspective

- The need to keep on reminding RDRL group that things are different in Wales to England!
 - E.g. “Resourcing and accessing a NGRL (national genomics research library) and secure data environment” not yet true for Wales
 - Rare disease clinical genomics data not easily searchable
 - Is “Investing strategically in research infrastructure” true for Wales?

Gaps identified from SWOT analysis (annex 1)	Existing investments (England) G = Strategic level investment A = non-strategic level investment or non-specific R = gap in investment	Existing investments (Wales)
Incentivising cross-sector collaborative research partnerships		
Maximising impact of rare disease research by identifying multiple impact routes (e.g. rare diseases with overlapping needs)		
Improving patient access to clinical trials, including recruitment by HCPs		
Investing strategically in research infrastructure		?
Addressing health inequalities		Issues of geography?
Resourcing and accessing a national genomic research library and secure data environment		Not available yet
Building on UK strength in mature patient advocacy groups		?
Building best practice into care pathways through research, including identifying commonalities between rare conditions		Rare care centre? SWAN (syndromes without a name) clinic?
Increasing access to molecular diagnoses		
Widening access to research and treatment opportunities		
Supporting research on ultra rare conditions ('long tail')		
Mapping of rare disease prevalence to research funding		

Research continues...

- HCRW champions “One Wales” for clinical research
 - Easy for NIHR clinical research formerly under the Genetics NSG
 - One AWMGS (All Wales Medical Genomics Service) as primary recruiter
- How could other Welsh rare disease researchers help each other?

CF: The journey to Improved Survival

Cystic Fibrosis Trust



64



Age (years)

40
35
30
25
20
15
10
5
0

1930

1940

1950

1960

1970

1980

1990

2000

2010

2024

1st pathologic description

Sweat chloride test developed

Centre care

National Registry
CF gene identified

Neonatal screening

RCTs

HTS

Azithromycin

Inhaled Tobramycin
rhDNase

Inhaled colistin

antipseudomonal antibiotics

antistaphylococcal antibiotics

Airway clearance

Pancreatic Enzymes

Colobreathe

Bronchitol

TIP

AZLI

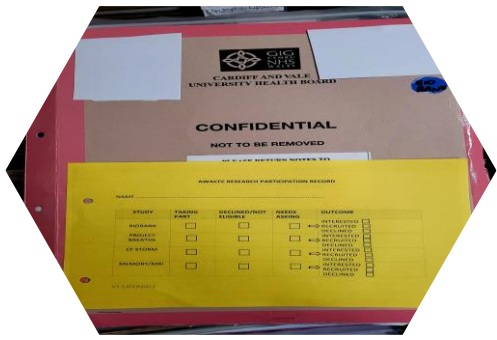
mRNA Gene therapy

Ivacaftor

Lum/Iva

Tez/Iva

ETI



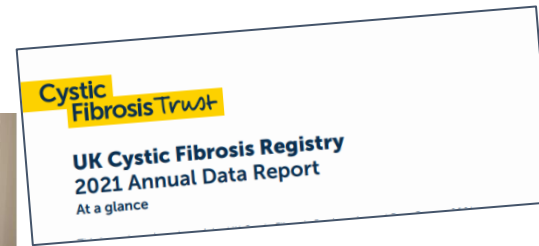
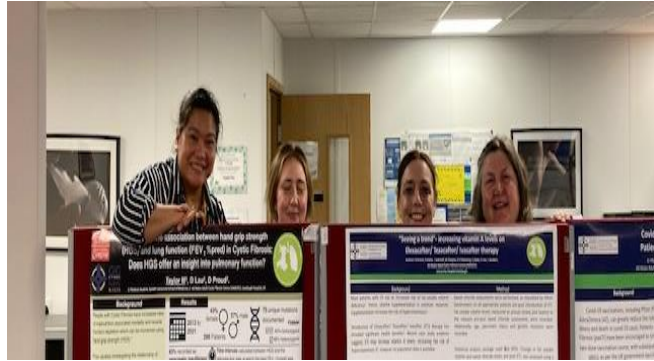
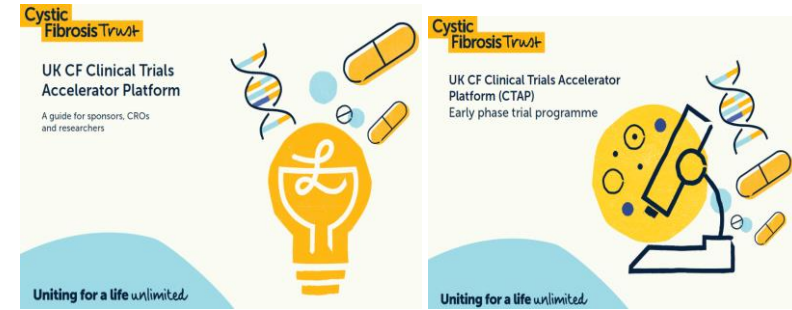
CF research model

- Embedding research into clinical care
- Engagement with charities
- Coordinated approach- networks
- PPI PPI PPI
- Power of registry data
- Power of biobanks

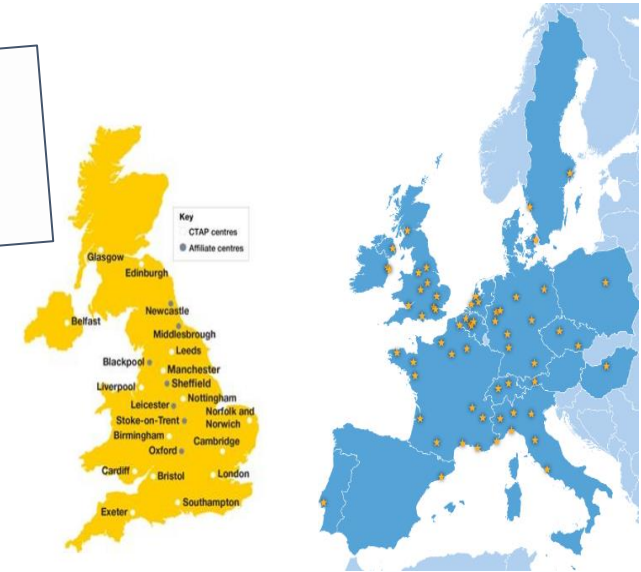


Trials Tracker

The Trials Tracker lists CF clinical trials recruiting in the UK. You can use the filters to find trials based on the trial status, location, etc.

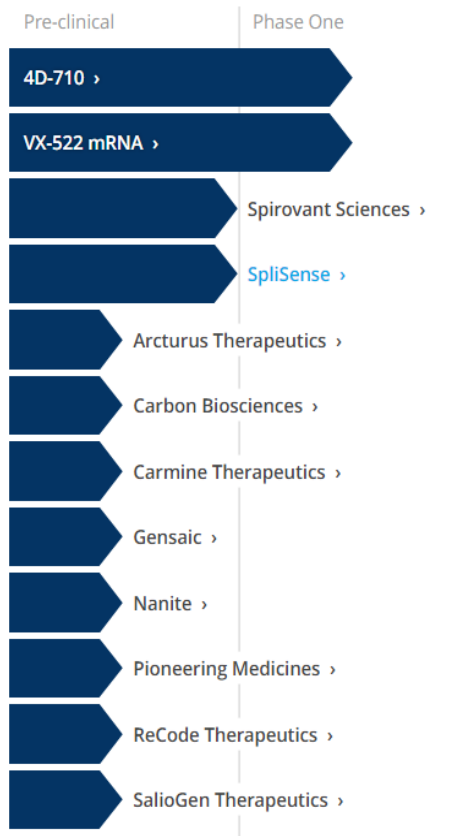


ECFS PATIENT REGISTRY

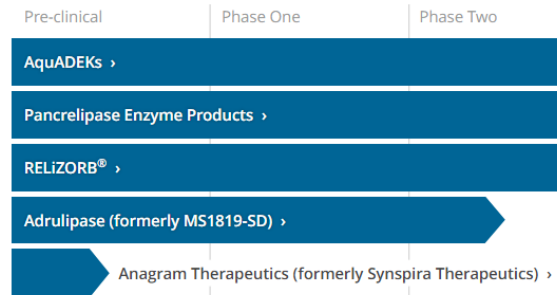


Genetic Therapy [Learn more >](#)

Gene therapy, RNA therapy, and antisense oligonucleotide

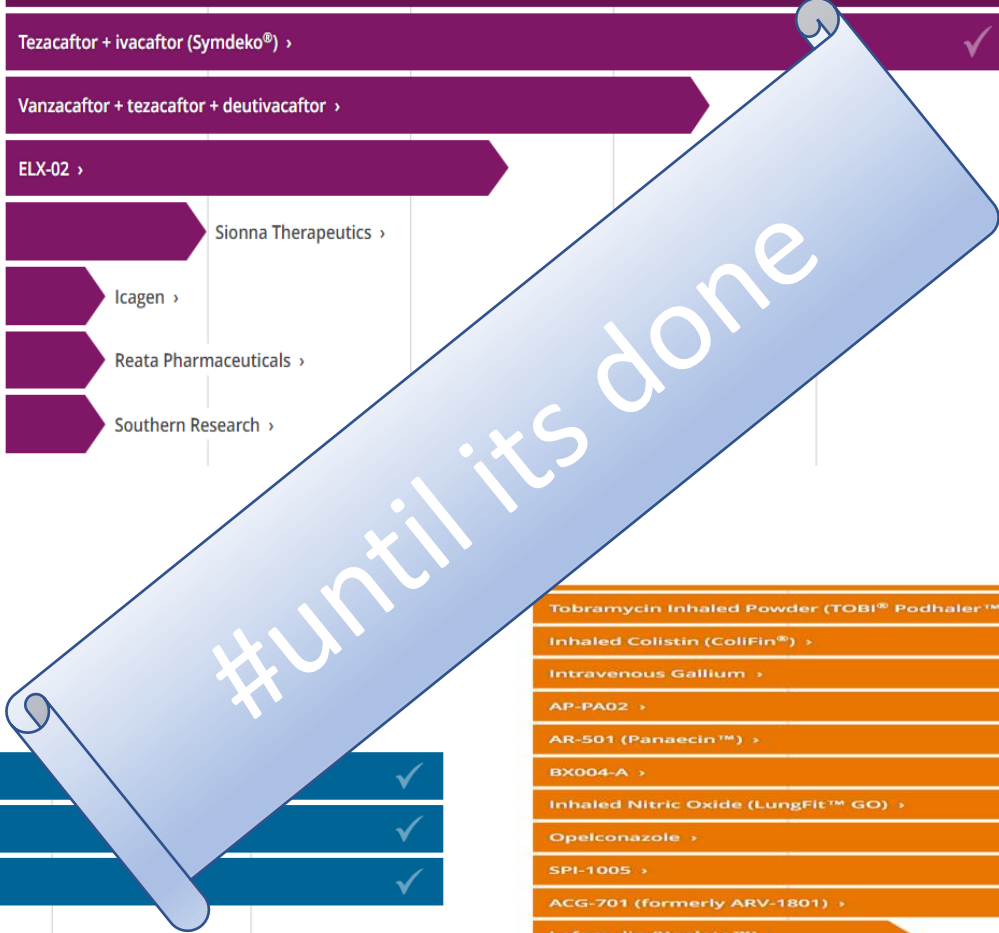
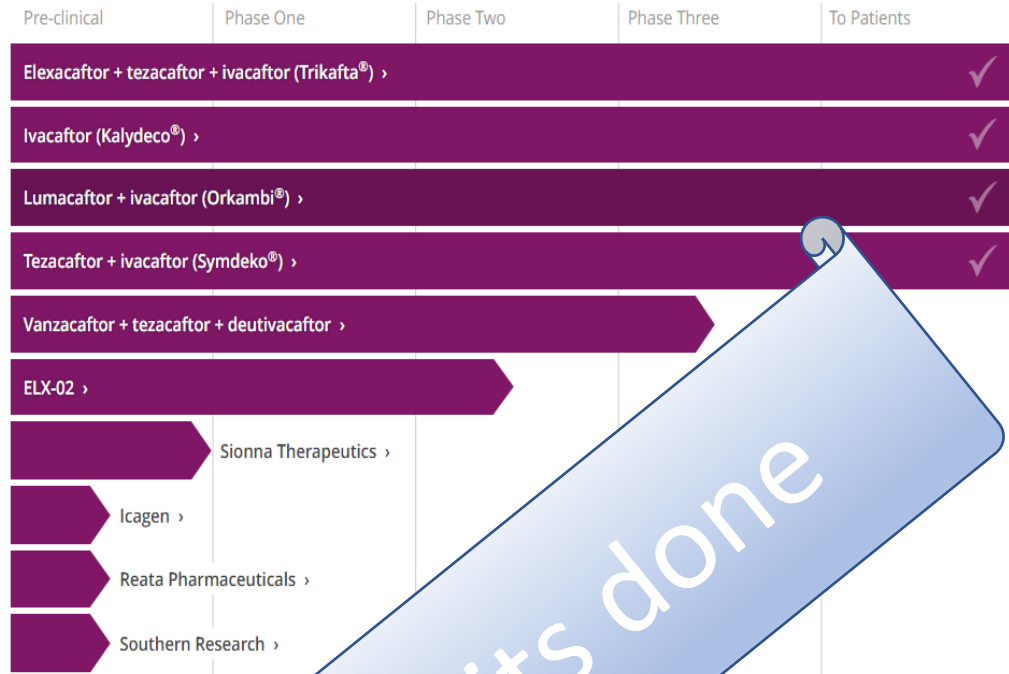


Nutritional-GI [Learn more >](#)

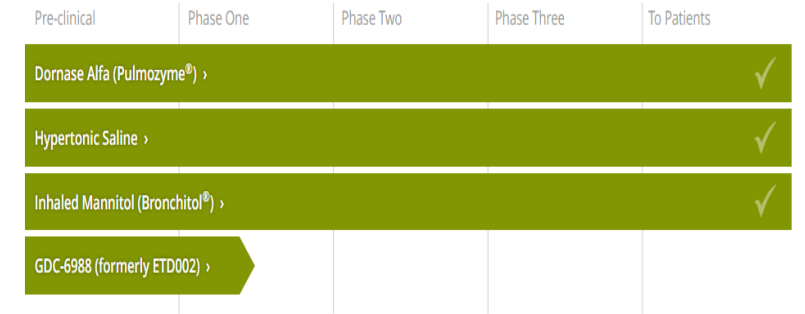


Restore CFTR Protein [Learn more >](#)

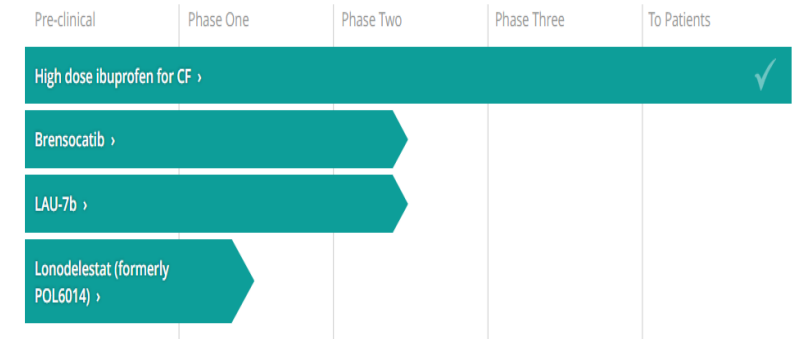
Modulators and nonsense readthrough therapies



Mucociliary Clearance [Learn more >](#)



Anti-Inflammatory [Learn more >](#)



Life Sciences Hub Wales

Liz Rees, Funding Programme Lead



What we do

We are an **arm's length body** of Welsh Government

We exist to catalyse **innovation** and **collaboration** between **industry, health, social care, and academia**

Our strategic aims:

- Improve health and wellbeing outcomes for people in Wales
- Improve efficiency and value within the health and social care system
- Drive economic development through business growth and jobs



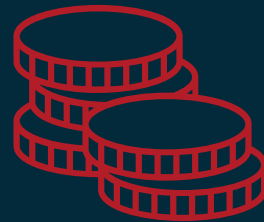
Our Delivery Teams



**Sector
Intelligence**



**Programme
Delivery**



**Economic
Development**



Partnerships



**Marketing &
Communications**



Our Current Priorities:

Digital

**Precision
Medicine**

Cancer

Funding Support

- Signposting to appropriate funding schemes
 - Funding web page
 - Research Connect database
- Bid Support
 - Eligibility Assessment
 - Staggered support for bid development
 - Bid review
- Networking for collaborative bids
 - Proactive consortia development
 - Connecting with funders UK-wide
 - Neutral broker



Funding Programmes

- UKRI (MRC, Innovate UK)
- NIHR
- SBRI & Challenge (signposting only)
- Welsh Government e.g. SMART FIS
- Health and Care Research Wales
- Charitable bodies e.g. Cancer Research UK, Moondance



Innovation Directory

Our Innovation Directory offers users essential details about organisations in Wales actively involved in the advancement of health and social care innovation. The directory currently lists:

- Life sciences companies based in Wales
- Health organisations – HBs, Trusts, RPBs etc.
- Innovation support organisations
- Universities & Intensive Learning Academies

<https://lshubwales.com/innovation-directory>





Hwb Gwyddorau Bywyd Cymru
Life Sciences Hub Wales

Any questions?

Join our community

in @lshubwales

X @lshubwales

✉ hello@lshubwales.com

lshubwales.com